ARGININOSUCCINIC ACIDURIA (ASA)
ASA is a urea cycle disorder resulting from a defect in the enzyme ASA lyase that converts ammonia to urea. This enzyme deficiency can present acutely in the newborn period with the elevated blood ammonia levels, seizures, failure to thrive, lethargy, and coma. Later signs include mental retardation.

Screening Method:
ASA and Citrulline levels are measured using a Tandem Mass Spectrometry method.

Treatment:
A low protein diet with arginine supplementation and patients must avoid fasting during illness.

Incidence:
1:60,000 births

CITRULLINEMIA (CIT)
Citrullinemia results from a deficiency of the enzyme Argininosuccinate Synthetase. This defect produces increased blood ammonia levels and are generally well for the first 24 –72 hours but then demonstrate lethargy, poor feeding, vomiting, respiratory distress, seizures, coma, and then death if not treated.

Screening Method:
Citrulline levels are measured using a Tandem Mass Spectrometry method.

Treatment:
Aggressive hemodialysis to clear ammonia from the blood. Maintenance treatment consists of a protein- restricted diet, medications to clear ammonia, and arginine supplementation.

Incidence:
1: 60,000 births.

HOMOCYSTINURIA (HCY)
Classical homocystinuria is due to deficiency of the enzyme Cystathiorine B-synthase (CBS). Methionine from ingested protein can not be converted to homocysteine due to deficiency of CBS resulting in elevated concentration of methionine and homocystiene in blood and urine of affected infants. Clinical problems include blood clots, seizures, optic lens dislocation, developmental and mental retardation.
Screening Method:
Methionine levels are measured using a Tandem Mass Spectrometry method.

Treatment:
Approximately 50% of patients respond to large doses of vitamin B6. Non responsive patients are treated with a methionine – restricted, cystine supplemental diet.

Incidence:
1: 300,000 births

MAPLE SYRUP URINE DISEASE (MSUD)
MSUD is caused by a deficiency in ability to metabolize the amino acids leucine, isoleucine, and valine (branch chain amino acids) in foods containing protein. This disorder presents in infants with vomiting, failure to thrive, lethargy, and a maple syrup odor to urine and ear wax. If untreated it will progress to irreversible mental retardation, irritability, coma and possibly death.

Screening Method:
Leucine and Valine levels are measured using a Tandem Mass Spectrometry method.

Treatment:
Dietary management with decreased leucine in the diet and limited isoleucine and valine. Aggressive management of acute metabolic acidosis.

Incidence:
1:250,000 births.

PHENYLKETONURIA (PKU)
Classical PKU is a metabolic disorder resulting in the Deficient production of the liver enzyme phenylalanine hydroxylase. This deficiency prevents the breakdown of phenylalanine, an amino acid present in all foods containing protein. The build-up of phenylalanine prevents the brain from developing and results in mental retardation.

Screening Method:
Phenylalanine levels are measured using a Tandem Mass Spectrometry method.

Treatment:
Special formula that provides nutrients needed for growth but restricts phenylalanine intake.
Incidence:
1: 14,000 births

TYROSINEMIA (TYR)
Tyrosinemia is a result of an enzyme deficiency that metabolizes the amino acid to tyrosine from ingested protein. Tyrosinemia I is usually a symptomatic and if untreated will cause liver disease and death from liver failure. Tyrosinemia II will cause corneal (eye) lesions, hyperkeratosis of the skin, and in some cases mental retardation.

Screening Method:
Tyrosine levels are measured using a Tandem Mass Spectrometry method.

Treatment:
Tyrosinemia Type I-Nitisinone (NTBC)
Tyrosinemia Type II- Tyrosine restricted diet

Incidence:
1: 63,000 births